Case reports

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A lethal short rib syndrome without polydactyly

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SUMMARY A female infant is described with a lethal short rib syndrome, similar to a form of short rib-polydactyly syndrome but without polydactyly. It is felt that this infant has the same condition as that described by Beemer *et al.*¹

Syndromes including short ribs represent a heterogeneous group of disorders. A subgroup consists of those conditions which are or can be lethal at or

before birth and these include thanatophoric dysplasia, achondrogenesis, Jeune syndrome, and the short rib-polydactyly syndromes. Beemer *et al.*¹ have described two patients with short ribs and early neonatal death. Although these infants showed similarities to some types of short rib-polydactyly syndrome, polydactyly was absent and it was suggested that they represented a new entity. A further female is described here showing greater similarity to the cases of Beemer *et al.*¹ than to any reported form of short rib-polydactyly syndrome.

FIG 1 AP photograph of the affected infant. Note relative macrocephaly, midline cleft upper lip, short limbs, and narrow chest.

FIG 2 AP radiograph of affected infant. Note short horizontal ribs, highly placed clavicles, small scapulae and ilia, short tubular bones with bowing of radius and ulna, and minimal metaphyseal irregularity or spiking.

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Case report

The female infant was the child of non-consanguineous Caucasian parents. The mother had had one previous early miscarriage and there was one normal female sib. Delivery was by caesarian section at 38½ weeks because of polyhydramnios and a large head. At birth the baby weighed 3.5 kg, length 44 cm (<3rd centile), and head circumference 38 cm (>98 centile). The limbs were markedly short with an arm span of 27 cm and a pubis to heel length of 13 cm. There was midline cleft upper lip and cleft palate with a very narrow thorax (fig 1). The digits were short, but there was no polydactyly. Radiographs showed very short, horizontal ribs, highly placed clavicles, small scapulae, small ilia, short tubular bones with bowing of the radius and ulna, and minimal metaphyseal irregularity and spiking (fig 2). Chromosomal analysis showed a normal female karyotype. Unfortunately cartilage histology was not examined and necropsy was not carried out.

Discussion

Although a diagnosis of one of the short rib-polydactyly syndromes manifesting without polydactyly must be considered here, this case is very similar to two unrelated cases, one born to a consanguineous couple, described by Beemer *et al.*¹ These authors did consider the possibility of their cases being examples of a type of short rib-polydactyly, particularly type II (Majewski); however the radiological features were not consistent with this condition. In short rib-polydactyly syndromes I (Saldino-Noonan) and III (Verma-Naumoff), metaphyseal irregularity and spiking is a marked feature.² It was not present in the case described here, apart from possible mild irregularity of the distal end of the femora. Short rib-polydactyly syndrome type II (Majewski) characteristically manifests with hypoplastic, oval shaped

TABLE Common features in three reported cases.

	Present case	Beemer <i>et al.</i> ¹	
		Case 1	Case 2
Macrocephaly	+	+	+
Flat face	+	+	+
Midline cleft lip	+	+	+
Cleft palate	+	+	—
Ascites	—	+	+
Omphalocele	—	+	+
Narrow thorax	+	+	+
Short ribs	+	+	+
Highly placed clavicles	+	+	+
Small scapulae	+	+	+
Small ilia	+	+	+
Short tubular bones	+	+	+
Bowing radius/ulna	+	+	+
Metaphyseal irregularity/spiking	+/-	—	—
Family history	Isolated case	Parents consanguineous, affected sib diagnosed prenatally by ultrasound	Isolated case

tibiae,³ quite unlike the well formed tibiae seen in the present case. Thus, it seems possible that a separate short rib syndrome is being described. The common features are shown in the table. Inheritance is assumed to be autosomal recessive, because of the sib pair reported by Beemer *et al.*¹

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Intrauterine death in megacystis-microcolon-intestinal hypoperistalsis syndrome

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SUMMARY Megacystis-microcolon-intestinal hypoperistalsis syndrome is an uncommon condition, possibly inherited as an autosomal recessive trait. This report describes an affected sib pair with intrauterine death of one of the sibs.

Winter and Knowles¹ recently described the occurrence of megacystis-microcolon-intestinal hypoperistalsis syndrome in two female sibs who were the offspring of first cousin parents, suggesting the possibility that this disorder has an autosomal recessive aetiology. Few cases of this condition, characterised by hypoperistalsis of the bowel and dysfunctional bladder contractility, have been reported and only three other possibly affected sib pairs have been noted.²⁻⁴ This report describes